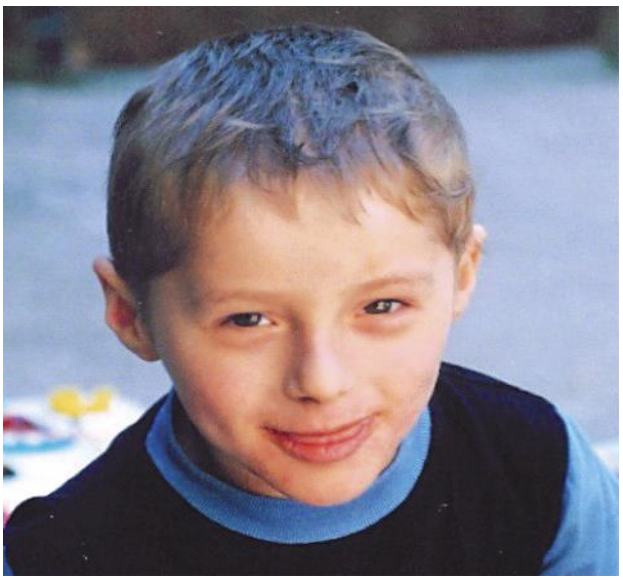
# Killian Association





## Syndrome of Angelman

The Syndrome of Angelman

The syndrome of Angelman is a rare genetic disease stemming from an abnormality on the chromosome 15 discovered in 1965 by the British paediatrician Harry Angelman. First estimated at 1/100000 we think today that the disease concerns 1 child for 10000 to 20000 births. This evolution shows progress of the science in the knowledge of the syndrome of Angelman.

*Several clinical signs are observed*:

**Constant signs**: a delay of the driving development, a severe mental delay, an almost absence of language, a stiff and jerky walk, a tendency to the easy laughter in situations which do not justify it. They are hyperactive children with periods of hyperexcitability being translated by characteristic beatings of the forearm.

**Frequent signs** in more than 80 % of the cases: epileptic activity beginning generally before three years, a plan of the particular electroencephalogram or the myoclonies.

**Signs associated** in more than 50 % of the cases: particular features of the face (wide mouth with spread superior teeth and thin superior lip, pointed chin and the back of the head flat, dribble, squint, hypo-pigmentation of the skin, hair or eyes, scoliosis and obesity in the adolescence. To note that disorders of the sleep are often observed.

This document was realized, financed and distributed by the Killian Association ( France ) with the aim of making the professionals of various countries sensitive to the specificities of the syndrome of Angelman. The Killian association consists of a group of friends united around a child. This association wants its action to be profitable for most large number. We try to make the syndrome of Angelman better known but we have no vocation to gather together families, it is necessary for it to contact the already existing national associations. To contact us: www.associationkillian.org

## The genetics of the syndrome of Angelman

The abnormalities are observed on the region 15q11q12 of the chromosome 15. Here are the main three abnormalities observed :

Microdélétion: loss of the region 15 q11q12 of the chromosome 15 of maternal origin in approximately 66 % of the cases with a hereditary risk of transmission lower than 1 %.

Disomie - parental: both chromosomes 15 result from the father in approximately 5 % of the cases with a hereditary risk of transmission lower than 1 %.

Mutation of the gene UBE3a: in approximately 10 % of the cases with a risk of hereditary transmission from 1 to 50 %.

There is approximately 10 % of the cases in which no abnormality is identified to this day.

## Treatment

No treatment is at present known. Only certain symptoms of the disease can be treated such the epilepsy, the disorders of the sleep, feeding or the eye disorders.

It is of the responsibility of all the concerned actors (scientists, doctors, parents, associations) to collaborate so that the information circulates in order to increase the number of detected patients and improve their coverage(care).

## Coverage(Care) / Care taking of

It is necessary to envisage a premature coverage(care) to increase at most the potential of the child and insure him most possible autonomy. This coverage(care) brings numerous actors. The **Physiotherapist** is often the first one request to help the nourisson to sit down then to help the child to walk. "Angelman" children walk generally later than the others.

This specialist also insures to the patient a correct muscle structure to avoid the appearance of scoliosis or the problems of retraction of tendons. A **psychomotricien** can come in complement to help the child in the field of the fine motricity. Afterward the **ortophonist** intervenes in the prevention or the reeducation of the dribble. It is a question of maintaining a good muscular tonic effect at the level of the jaw. This professional also contributes to maintain his patient in the envy to communicate. The reproduction of different stimulations opens the child to the world which surrounds him. They are particularly sociable children who are very attracted by water, music, objects which turn (wheels, tops, balloons).

## Interests of the detection

This interest is double! At first it is a question of offering to the handicaped person the coverage (care) which corresponds best to the met difficulty. Secondly it is to be hoped that the increase of the number of detected persons catch the attention in order to stimulate the research and the interest of the pharmaceutical industry.

## he objectives of this document

The Killian association, according to its status wishes the syndrome of Angelman to be better known. We hope that this brochure helps in the detection of new cases in countries where the disease is little known even absent. It is important to detect the " Angelman "children to propose them the appropriate coverage(care) to allow them to escape from a restrictive psychiatric label. Through this support we want to give a first approach of the syndrome of Angelman but we do not claim to be able to substitute ourselves for the scientists who are the only one qualified to develop more precise information. The Killian association is a small structure having a limited budget, so we are forced to limit the edition of this document to 1000 copies by country, if you join our project, you can make this brochure circulate with your collaborators. We would be happy that you inform us of positive effects of this document on the web site of our association: www.associationKillian.org. In some months we shall propose on our Web site a translation of this working tool in several languages.